



CST3 gene

cystatin C

Normal Function

The *CST3* gene provides instructions for making a protein called cystatin C. This protein is part of a family of proteins called cysteine protease inhibitors that help control several types of chemical reactions by blocking (inhibiting) the activity of certain enzymes. Cystatin C inhibits the activity of enzymes called cathepsins that cut apart other proteins in order to break them down.

Cystatin C is found in biological fluids, such as blood. Its levels are especially high in the fluid that surrounds and protects the brain and spinal cord (the cerebrospinal fluid or CSF).

Health Conditions Related to Genetic Changes

age-related macular degeneration

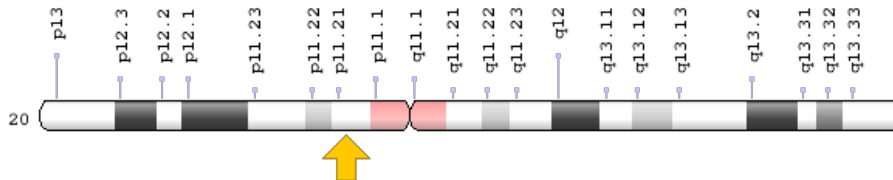
hereditary cerebral amyloid angiopathy

At least one mutation in the *CST3* gene has been found to cause hereditary cerebral amyloid angiopathy, a condition characterized by stroke and a decline in intellectual function (dementia), which begins in mid-adulthood. The *CST3* gene mutation that has been identified causes a form of hereditary cerebral amyloid angiopathy known as the Icelandic type. This mutation replaces the protein building block (amino acid) leucine with the amino acid glutamine at position 68 in the cystatin C protein (written as Leu68Gln or L68Q). This abnormal cystatin C protein is less stable and is more prone to cluster together (aggregate) than the normal protein. The aggregated protein forms clumps called amyloid deposits that accumulate in the blood vessel walls primarily in the brain, but also in blood vessels in other areas of the body such as the skin, spleen, and lymph nodes. The accumulation of these amyloid deposits, known as plaques, does not appear to have any health effects outside of the brain. In the brain, the amyloid plaques replace the muscle fibers and elastic fibers that give blood vessels flexibility, causing them to become weak and prone to breakage. Such a break in the brain causes bleeding (hemorrhagic stroke), which can lead to brain damage and dementia.

Chromosomal Location

Cytogenetic Location: 20p11.21, which is the short (p) arm of chromosome 20 at position 11.21

Molecular Location: base pairs 23,627,897 to 23,638,048 on chromosome 20 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- cystatin-3
- cystatin 3
- cystatin-C
- cystatin-C precursor
- CYTC_HUMAN
- gamma-trace
- neuroendocrine basic polypeptide
- post-gamma-globulin

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CST3%5BTIAB%5D%29+OR+%28cystatin+C%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

OMIM

- CYSTATIN 3
<http://omim.org/entry/604312>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_CST3.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=CST3%5Bgene%5D>
- HGNC Gene Family: Cystatins, type 2
<http://www.genenames.org/cgi-bin/genefamilies/set/965>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2475
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/1471>
- UniProt
<http://www.uniprot.org/uniprot/P01034>

Sources for This Summary

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<https://ghr.nlm.nih.gov/gene/CST3>

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